REN-related kidney disease

REN-related kidney disease is an inherited condition that affects kidney function. This condition causes slowly progressive kidney disease that usually becomes apparent during childhood. As this condition progresses, the kidneys become less able to filter fluids and waste products from the body, resulting in kidney failure. Individuals with *REN*-related kidney disease typically require dialysis (to remove wastes from the blood) or a kidney transplant between ages 40 and 70.

People with *REN*-related kidney disease sometimes have low blood pressure. They may also have mildly increased levels of potassium in their blood (hyperkalemia). In childhood, people with *REN*-related kidney disease develop a shortage of red blood cells (anemia), which can cause pale skin, weakness, and fatigue. In this disorder, anemia is usually mild and begins to improve during adolescence.

Many individuals with this condition develop high blood levels of a waste product called uric acid. Normally, the kidneys remove uric acid from the blood and transfer it to urine so it can be excreted from the body. In *REN*-related kidney disease, the kidneys are unable to remove uric acid from the blood effectively. A buildup of uric acid can cause gout, which is a form of arthritis resulting from uric acid crystals in the joints. Individuals with *REN*-related kidney disease may begin to experience the signs and symptoms of gout during their twenties.

Frequency

REN-related kidney disease is a rare condition. At least three families with this condition have been identified.

Genetic Changes

Mutations in the *REN* gene cause *REN*-related kidney disease. This gene provides instructions for making a protein called renin that is produced in the kidneys. Renin plays an important role in regulating blood pressure and water levels in the body.

Mutations in the *REN* gene that cause *REN*-related kidney disease result in the production of an abnormal protein that is toxic to the cells that normally produce renin. These kidney cells gradually die off, which causes progressive kidney disease.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Familial juvenile hyperuricemic nephropathy 2

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Hyperuricemic nephropathy, familial juvenile, 2
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2751310/

Other Diagnosis and Management Resources

- GeneReview: Autosomal Dominant Tubulointerstitial Kidney Disease, REN-Related
  https://www.ncbi.nlm.nih.gov/books/NBK53700
- MedlinePlus Encyclopedia: Hyperkalemia
  https://medlineplus.gov/ency/article/001179.htm
- MedlinePlus Encyclopedia: Renin
  https://medlineplus.gov/ency/article/003698.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Hyperkalemia
  https://medlineplus.gov/ency/article/001179.htm
- Encyclopedia: Renin
  https://medlineplus.gov/ency/article/003698.htm
- Health Topic: Anemia
  https://medlineplus.gov/anemia.html

- Health Topic: Gout
  https://medlineplus.gov/gout.html
- Health Topic: Kidney Diseases
  https://medlineplus.gov/kidneydiseases.html

Additional NIH Resources

- National Institute of Arthritis and Musculoskeletal and Skin Diseases: Gout
  https://www.niams.nih.gov/Health_Info/Gout/
- National Institute of Diabetes and Digestive and Kidney Diseases: The Kidneys and How They Work
  https://www.niddk.nih.gov/health-information/kidney-disease/kidneys-how-they-work

Educational Resources

- MalaCards: ren-related kidney disease
  http://www.malacards.org/card/ren_related_kidney_disease
- Merck Manual Consumer Version: Gout
  http://www.merckmanuals.com/home/bone-joint-and-muscle-disorders/gout-and-pseudogout/gout
- Merck Manual Consumer Version: The Body's Control of Blood Pressure
  http://www.merckmanuals.com/home/heart-and-blood-vessel-disorders/high-blood-pressure/high-blood-pressure
- Wake Forest University Baptist Medical Center: Inherited Kidney Disease
  http://www.wakehealth.edu/Nephrology/Inherited-Kidney-Disease.htm

Patient Support and Advocacy Resources

- National Kidney Foundation
  https://www.kidney.org/

GeneReviews

- Autosomal Dominant Tubulointerstitial Kidney Disease, REN-Related
  https://www.ncbi.nlm.nih.gov/books/NBK53700

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28familial+juvenile+hyperuricemic+nephropathy+2%29+OR+%28%28REN%29+AND+%28gene%29%29+AND+%28renin%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D

OMIM

- HYPERURICEMIC NEPHROPATHY, FAMILIAL JUVENILE, 2
  http://omim.org/entry/613092

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